



Patient ID SA00108233	Patient Name TESTINGRNV, SAMPLEREPORT	Birth Date 1982-06-29	Gender F	Age 35
Order Number SA00108233	Client Order Number SA00108233	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 26 Jun 2018 10:00		

Hepatosplenomegaly Panel, P

Interpretation (HSMP)

POSITIVE In this sample, the oxysterol profile and the lysosphingomyelin509 are suggestive of a biochemical diagnosis of Niemann-Pick disease type A/B.

ADDITIONAL INFORMATION

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

Cholestane-3beta,5alpha,6beta-triol

 0.071 nmol/mL
High

MCR
Reference Value
≤ 0.070

7-Ketocholesterol

 0.101 nmol/mL
High

MCR
Reference Value
≤ 0.100

Lyso-sphingomyelin

 0.101 nmol/mL
High

MCR
Reference Value
≤ 0.100

Glucopsychosine

 0.004 nmol/mL
High

MCR
Reference Value
≤ 0.003

7a-hydroxy-4-cholesten-3-one

 0.301 nmol/mL
High

MCR
Reference Value
≤ 0.300

7a,12a-dihydroxycholest-4-en-3-one

 0.101 nmol/mL
High

MCR
Reference Value
≤ 0.100

Reviewed By

Amy Breitlow

MCR

Received: 27 Jun 2018 11:30

Reported: 27 Jun 2018 12:13

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292